BDNF gene

brain derived neurotrophic factor

Normal Function

The *BDNF* gene provides instructions for making a protein found in the brain and spinal cord called brain-derived neurotrophic factor. This protein promotes the survival of nerve cells (neurons) by playing a role in the growth, maturation (differentiation), and maintenance of these cells. In the brain, the BDNF protein is active at the connections between nerve cells (synapses), where cell-to-cell communication occurs. The synapses can change and adapt over time in response to experience, a characteristic called synaptic plasticity. The BDNF protein helps regulate synaptic plasticity, which is important for learning and memory.

The BDNF protein is found in regions of the brain that control eating, drinking, and body weight; the protein likely contributes to the management of these functions.

Health Conditions Related to Genetic Changes

WAGR syndrome

The *BDNF* gene is located in a region of chromosome 11 that is often deleted in a condition known as WAGRO syndrome. This condition is a variant of WAGR syndrome, which is a disorder that affects many body systems and is named for its main features: a childhood kidney cancer known as Wilms tumor, an eye problem called anirida, genitourinary anomalies, and intellectual disability (formerly referred to as mental retardation). WAGRO syndrome also includes obesity. The deletions that cause WAGRO syndrome remove many genes from one copy of chromosome 11, including part or all of the *BDNF* gene. The loss of this gene is responsible for weight gain that begins in childhood in people with WAGRO syndrome.

People with WAGRO syndrome may be at greater risk of neurological problems such as intellectual disability and a developmental disorder called autism that affects communication and social interaction than those with WAGR syndrome. It is unclear whether this increased risk is due to the loss of the *BDNF* gene or other nearby genes.

other disorders

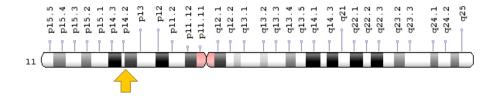
Certain common genetic variations (polymorphisms) in the *BDNF* gene have been associated with an increased risk of developing psychiatric disorders such as bipolar disorder, anxiety, and eating disorders.

Most studies have focused on the effects of a particular polymorphism in the *BDNF* gene. This variation alters a single protein building block (amino acid) in the protein, replacing the amino acid valine with the amino acid methionine at position 66 (written at Val66Met or V66M). This change impairs the protein's ability to function. Many studies report an association between the Val66Met polymorphism and psychiatric disorders; however, some studies have not supported these findings. It is unclear how changes in the *BDNF* gene are related to these disorders. A large number of genetic and environmental factors, most of which remain unknown, likely determine the risk of developing these complex conditions.

Chromosomal Location

Cytogenetic Location: 11p14.1, which is the short (p) arm of chromosome 11 at position 14.1

Molecular Location: base pairs 27,654,893 to 27,722,058 on chromosome 11 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- abrineurin
- ANON2
- BDNF_HUMAN
- brain-derived neurotrophic factor
- BULN2
- neurotrophin

Additional Information & Resources

Scientific Articles on PubMed

PubMed

https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28BDNF%5BTI%5D%29+OR+%28brain-derived+neurotrophic+factor%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D

OMIM

- BRAIN-DERIVED NEUROTROPHIC FACTOR http://omim.org/entry/113505
- BULIMIA NERVOSA, SUSCEPTIBILITY TO, 2 http://omim.org/entry/610269

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology http://atlasgeneticsoncology.org/Genes/GC_BDNF.html
- ClinVar https://www.ncbi.nlm.nih.gov/clinvar?term=BDNF%5Bgene%5D
- HGNC Gene Family: Endogenous ligands http://www.genenames.org/cgi-bin/genefamilies/set/542
- HGNC Gene Symbol Report http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/ hgnc_data.php&hgnc_id=1033
- NCBI Gene https://www.ncbi.nlm.nih.gov/gene/627
- UniProt http://www.uniprot.org/uniprot/P23560

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